



Think Fabry disease and the heart!

Cardiac complications are the leading cause of mortality in Fabry disease.¹

What is Fabry disease?

Fabry disease is an x-linked inherited lysosomal storage disorder that results from the buildup of globotriaosylceramide and globotriaosylsphingosine, in the body's cells. Beginning in childhood, this buildup causes signs and symptoms that can affect multiple organs of the body. Fabry disease also involves potentially life-threatening complications such as progressive kidney damage, heart attack and stroke. Some affected individuals have milder forms of the disorder that appear later in life and affect only the heart or kidneys.²

Consider screening for low levels of alpha galactosidase A enzyme for patients diagnosed with:

- LVH or hypertrophic cardiomyopathy without known causes
- Patients ≥ 20 years of age with LVH wall thickness ≥ 12 mm
- Cardiac arrhythmias
- Myocardial infarction or congestive heart failure without known causes
- Family history of Fabry disease

If Fabry disease is suspected:

Cardiologists should consider **referring** to and **engaging** your local expert **Fabry centre** for conclusive diagnosis.

If you suspect Fabry disease, please refer to the list of Australian Fabry Clinics listed on the reverse.



Australian Fabry Clinic locations:

VIC

Royal Melbourne Hospital

Nephrology Dept

Contact:

A/Prof Kathy Nicholls
Elizabeth Centra

Tel: 03 9342 7143/6460

QLD

Royal Brisbane & Womans Hospital

Internal Med & Aged Care

Contact:

A/Prof Charles Denaro

Tel: 07 3646 5385

NSW

Westmead Hospital

Genetics & Metabolic Clinic

Contact:

Dr Yusof Rahman
Kerry Devine

Tel: 02 8890 9780

Email: WSLHD-Geneticmedicine-Westmead@health.nsw.gov.au

SA

Royal Adelaide Hospital

Endocrine & Metabolic Dept

Contact:

Prof Ian Chapman
Carmen Bishoff

Tel: 7074 2674

WA

Royal Perth Hospital

Nephrology Dept

Contact:

Prof Mark Thomas

Tel: 08 9224 2550

References:

1. Germain DP. Fabry disease. Orphanet J Rare Dis.2010;5:30
2. <https://ghr.nlm.nih.gov/condition/fabry-disease>.

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